

Listing of Claims

1. (Currently amended) A method of using statistical analysis of genetic data from an inbred population to determine likely genetic regions for a recessive genetic disease or trait, comprising the steps of:

obtaining actual genotype data from members of an inbred population, wherein said members are selected from one or both of: people affected with a genetic disease or trait in said inbred population and parents of people affected with said genetic disease or trait in said inbred population;

obtaining estimated genotype data for said inbred population; and

analyzing the actual and estimated genotype data to find one or both of a region in genomes of the affected people and/or a region in genomes of parents of the affected people, wherein said region in genomes of the affected people and said region in genomes of parents of the affected people include includes markers exhibiting particular homozygous pairs of alleles more frequently than would occur randomly and said step of analyzing is performed using a computing device, and wherein said step of analyzing comprises:

determining a set of scores for each of said markers in said actual and estimated genotype data relative to each person for which actual genotype data was determined, with the set of scores for each marker including at least first scores generated to determine probabilities of observing each marker given autozygosity with a founder and second scores generated to determine probabilities of observing each marker given absence of autozygosity with the founder;

merging the set of scores for each marker to produce a merged score for each marker, wherein said step of merging comprises:

computing for each of said markers a ratio of said first scores to said second scores,

wherein each merged score indicates at least in part a statistical distinction between whether said marker is autozygous and whether said marker is not autozygous;

examining the merged scores to determine one or more contiguous regions of markers by locating a statistically significant gap in sums of said

merged scores for non-overlapping contiguous regions of markers, wherein contiguous regions of markers having scores above the gap are determined to be said one or more contiguous regions of markers;

selecting from said one or more contiguous regions of markers at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait; and

storing said at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait to a computer-readable memory; and

sequencing DNA in said at least one contiguous region to identify the recessive allele associated with said genetic disease or trait.

2. (Previously presented) A method as in claim 1, wherein said inbred population is a relatively inbred population with a higher occurrence of said genetic disease or trait than a more general population.
3. (Previously presented) A method as in claim 2, wherein the particular homozygous pairs of alleles are autozygous alleles descended from a founder of said genetic disease or trait in said relatively inbred population.
4. (Previously presented) A method as in claim 3, wherein each merged score for a marker represents a comparison of a likelihood of observing said marker given that people with said genetic disease or trait are autozygous at said marker versus a likelihood of observing said marker given that alleles for said marker are independent of said genetic disease or trait.
5. (Previously presented) A method as in claim 4, wherein said marker receives a higher merged score from one form of homozygosity versus another form of homozygosity, with the form receiving said higher score being more likely to be associated with said genetic disease or trait.
6. (Currently amended) A method as in claim 5, wherein said merged scores are placed in

an array ordered by a chromosomal order of markers associated with the merged scores.

7. (Previously presented) A method as in claim 6, wherein identifying said at least one contiguous region further comprises determining a consecutive portion of said array that has the highest sum.
8. (Previously presented) A method as in claim 6, wherein identifying said at least one contiguous region further comprises computing all sums of a predetermined fixed number of adjacent elements in said array and comparing the sums.
9. (Canceled)
10. (Canceled)
11. (Currently amended) A method of analyzing actual and estimated genotype data, with the actual genotype data obtained for one or more affected people with the genetic disease or trait in an inbred population, for their parents, or for the affected people and their parents, and with the estimated genotype data obtained for said population, the method performed to find a region in genomes of the affected people or a region in genomes of parents of the affected people, wherein said region includes markers exhibiting particular homozygous pairs of alleles more frequently than would occur randomly, the method comprising:
 - determining a set of scores for each marker in said actual and estimated genotype data relative to each person for which actual genotype data was determined, with the set of scores for each marker including at least first scores generated to determine probabilities of observing each marker given autozygosity with ~~the~~ a founder and second scores generated to determine probabilities of observing each marker given absence of autozygosity with the founder;
 - merging the set of scores for each marker to produce a merged score for each marker, wherein said step of merging comprises:
 - computing for each of said markers a ratio of said first scores to said second scores,

wherein each merged score indicates at least in part a statistical distinction between whether said marker is autozygous and whether said marker is not autozygous;

examining the merged scores to determine one or more contiguous regions of markers by locating a statistically significant gap in sums of said merged scores for non-overlapping contiguous regions of markers, wherein contiguous regions of markers having scores above the gap are determined to be said one or more contiguous regions of markers;

selecting from said one or more contiguous regions of markers at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait; and

storing said at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait to a computer-readable memory; and

sequencing DNA in said at least one contiguous region to identify the recessive allele associated with said genetic disease or trait;

wherein said determining and merging steps are performed using a computing device.

12. (Previously presented) A method as in claim 11, wherein said population is a relatively inbred population with a higher occurrence of said genetic disease or trait than a more general population.

13. (Previously presented) A method as in claim 12, wherein the particular homozygous pairs of alleles are autozygous alleles descended from a founder of said genetic disease or trait in said relatively inbred population.

14. (Previously presented) A method as in claim 13, wherein each merged score for a marker represents a comparison of a likelihood of observing said marker given that people with said genetic disease or trait are autozygous at said marker versus a likelihood of observing said marker given that alleles for the marker are independent of said genetic

disease or trait.

15. (Previously presented) A method as in claim 14, wherein said marker receives a higher merged score from one form of homozygosity versus another form of homozygosity, with the form receiving said higher score being more likely to be associated with said genetic disease or trait.

16. (Currently amended) A method as in claim 15, wherein said merged scores are placed in an array ordered by a chromosomal order of markers associated with the merged scores.

17. (Previously presented) A method as in claim 16, wherein identifying said at least one contiguous region further comprises determining a consecutive portion of said array that has the highest sum.

18. (Previously presented) A method as in claim 16, wherein identifying said at least one contiguous region further comprises computing all sums of a predetermined fixed number of adjacent elements in said array and comparing the sums.

19. (Canceled)

20. (Canceled)

21. (Currently amended) An apparatus including:

a processor;

input and output interfaces; and

a memory storing instructions executable by the processor to analyze actual and estimated genotype data, with the actual genotype data obtained for one or more affected people with the genetic disease or trait in an inbred population, for their parents, or for the affected people and their parents, and with the estimated genotype data obtained for said population, the method performed to find a region in genomes of the affected people or a region in genomes of parents of the affected people, wherein said region includes markers

exhibiting particular homozygous pairs of alleles more frequently than would occur randomly, the instructions including steps of:

(a) determining a set of scores for each marker in said actual and estimated genotype data relative to each person for which actual genotype data was determined, with the set of scores for each marker including at least first scores generated to determine probabilities of observing each marker given autozygosity with a founder and second scores generated to determine probabilities of observing each marker given absence of autozygosity with the founder;

(b) merging the scores for each marker to produce a merged score for each marker, wherein said step of merging comprises:

computing for each of said markers a ratio of said first scores to said second scores, wherein each merged score indicates at least in part a statistical distinction between whether said marker is autozygous and whether said marker is not autozygous;

(c) examining the merged scores to determine one or more contiguous regions of markers by locating a statistically significant gap in sums of said merged scores for non-overlapping contiguous regions of markers, wherein contiguous regions of markers having scores above the gap are determined to be said one or more contiguous regions of markers;

(d) selecting from said one or more contiguous regions of markers at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait; and

(f) storing said at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait to a computer-readable memory; and

(g) sequencing DNA in said one or more contiguous regions of markers to identify the recessive allele associated with said genetic disease or trait.

22. (Previously presented) A method as in claim 1, further comprising the step of sequencing said at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait.

23. (Previously presented) A method as in claim 11, further comprising the step of sequencing said at least one contiguous region likely to contain a recessive allele associated with said genetic disease or trait.